Clinical Profile Of Atrial Septal Defects In children at Sohag University hospital
Mohamed Abdel Aal M.Bakheet, Safaa Husein Ali, Doaa Maghraby Bahig
Pediatric Department, Faculty of Medicine, Sohag University, Sohag Egypt.

Abstract:
Background: Cardiac defects are the most common type of birth defects. Atrial Septal Defect (ASD) is a common cardio-vascular malformation, it is caused by the spontaneous malformation of the interatrial septum resulting in one or more openings, commonly referred to as holes in the heart. Patients and methods: This study is a prospective hospital-based study involving all children at the age from 0 to 18 years diagnosed by echo-cardiography as having isolated atrial septal defect and admitted to pediatric department, intermediate care, and neonatal intensive care unit (NICU); also outpatient clinic at Sohag university hospital during the period from January 2012 to January 2013. Results: We studied 65 patients diagnosed by trans-thoracic echo-cardiography as isolated atrial septal defect (ASD), Their ages ranged from 5 days to 13 years with Mean ± SD (2.27 ± 2.8 years). The main presenting symptoms included; recurrent chest infections, respiratory distress, feeding difficulties, exertional dyspnea, failure to thrive, manifestations of heart failure, and permanent cyanosis. Echo findings demonstrated that the majority of cases had ASDII, followed by ASDI, then combined ASD, and less common type was sinus venosus ASD. Conclusion: The great majority of infants with ASDII are most often asymptomatic. Frequently, the condition is detected by a murmur on routine physical examination when they are school-aged children.
Keywords: Atrial Septal Defect, echo-cardiography, murmur.

Introduction:
Atrial Septal Defect (ASD) is a common cardiovascular malformation, it is caused by the spontaneous malformation of the interatrial septum resulting in one or more openings, commonly referred to as holes in the heart. It affects over 1 in 1000 live births, accounting for 10% of congenital heart defects (CHD). Hoffmann JI et al., 2002. Ostium secundum type (ASDII) is the most prevalent, representing approximately 85% of all ASDs (Jacobs JP et al., 2000). Incidence of ASD has increased due to improved detection with the spread of routine diagnostic echocardiography. (Markham LW et al., 2011).
Small - sized Atrial Septal Defects usually do not cause symptoms during infancy and early childhood especially those of secundum type & normal pulmonary vascular resistance. Many individuals may live their entire lives with small ASDs while remaining unaware that any abnormality exists. Bloch M, 2012, often they are not diagnosed until later in life, because the only manifestations are a soft murmur and possibly an abnormal second sound. Abraham M. Rudolph, 2009. However, Children with medium & large - sized defects may have poor physical development and frequently have both heights and weights in the lower percentiles (failure to thrive), recurrent chest infections, poor feeding……etc. Most children have normal exercise tolerance, but occasionally a child
Children with Atrial Septal Defects are at increased risk for several complications, such as endocarditis (if associated mitral valve insufficiency is present), except ASDII as no risk of endocarditis is expected with ASDII except 6 months after surgical or transcatheter closure, and respiratory tract infections, which are less well tolerated in children with atrial septal defects than in children without Atrial Septal Defects. Furthermore, children with clinically significant and untreated Atrial Septal Defects are at risk for various cardiac complications, including CHF, pulmonary hypertension, and arrhythmias. (Michael R Carr et al., 2012).

II. Complete clinical examinations focusing on:
General look, Anthropometric measurements: weight and height, Vital signs, Detailed general examinations, and Detailed cardiac examinations.

III. Presence or absence of complications.

IV. Investigations:
Routine laboratory investigations which include:
Complete blood picture, Blood culture: in suspected cases of infective endocarditis, CRP and blood gases: if indicated.
Imaging which includes:
Chest x-ray, Electrocardiography (ECG), Echocardiography (TTE): we use in the study trans-thoracic echo two dimensional, color Doppler and M-mode (7MHZ, 5MHZ transducers frequency)

V. Therapeutic modalities: at time of the study.

Statistical analysis:
Statistical analysis will be done using SPSS-16. Data will be expressed as means and SD.
Tests: S- student test & chi square test will be used. P-value < 0.05 is significant, > 0.05 is in-significant, and < 0.01 is highly significant.

Ethical consideration:
Approval of the Sohag faculty of medicine research ethical committee will be taken and informed consent from the parents will be obtained.

3- Results:
We studied [65] patients diagnosed by trans-thoracic echo-cardiography as isolated atrial septal defect (ASD), Their ages ranged from 5 days to 13
years with Mean ± SD (2.27 ± 2.8 years), as for gender distribution of our studied cases (25) were males (38.4%), while (40) were females (61.5%) with female predominance; male to female ratio was [1: 1.6], as regards to age at presentation, we found that most of our studied cases 32 (49.2%) presented during infancy, followed by 13 (20%) during neonatal period, then 9 (13.8%) during preschool age & 9 (13.8%) during school age, meanwhile 2 (3.1%) presented during adolescence; the less frequent age category. (Table 1)

As regard to residence, we found that the majority of children 45 (69.2%) belonged to rural areas, while 20 (30.8%) belonged to urban and semi-urban areas with a statistically significant difference between both (P-value = 0.01). We detected positive consanguinity among parents of 50 cases (76.9%), most of them 42 (64.6%) had 1st-degree consanguinity.

Figure (1)

Figure (2) shows the main presenting symptoms of cases with all types of ASD detected at time of presentation, we found that 31 patients (47.7%) were asymptomatic, and discovered accidentally during routine medical examination; of them 29 cases had ASDII while the other 2 cases had ASDI & sinus venous lesions respectively. 34 cases (52.3%) were symptomatic; of them 6 had primum ASD, 1 had sinus venous, while 3 cases had combined ASD. Symptoms included; recurrent chest infections in 24 (36.9%) the commonest, followed by respiratory distress in 21 (32.3%), feeding difficulties in 20 (30.8%), exertional dyspnea in 18 (27.7%), failure to thrive in 17 (26.2%), manifestations of heart failure in 17 (26.2%), and the less common was permanent cyanosis i.e Eisenmenger syndrome in 3 cases (4.6%) | 3 cases of primum ASD of them 1 case had down syndrome | those who developed severe PHT with subsequent reversal of the shunt direction. Thirty-one (31) patients were asymptomatic, of the 29 cases had ASDII (93.54%) while the other 2 cases had ASD primum & sinus venous lesions representing (3.23%) per each.

Table (2) shows echocardiographic findings in our study demonstrated that the majority of cases had ASDII in 53 (81.5%), followed by ASDI in 7 (10.8%), then combined ASD in 3 (4.6%), and the less common type was sinus venous ASD in 2 (3.1). Regarding size of the defect as measured by ECHO, it ranged from (1.5 - 23 mm) with mean ± SD (7.9 ± 4.7), small defects (2 – 5 mm) were detected in 26 cases (40%), medium-sized (5 - 8 mm) in 18 (27.7%), and large-sized defects (> 8mm) in 21 (32.3%) of cases. As for direction of the shunt; the vast majority of cases 62 (95.4%) had left-to-right shunt, while right-to-left shunt was found in 3 (4.6%) of patients those who developed severe PHT & Eisenmenger syndrome. As regard pulmonary blood pressure (PBP); 51 (78.5%) of cases were normal, while 14 (21.5%) had pulmonary hypertension (PHT). PBP in our patients ranged from (35 - 71 mmHg) with Mean ± SD (49.5 ± 13.06 mmHg). Moderate PHT (41 - 55 mm Hg) was detected in 7 (10.8%), severe (> 55 mm Hg) in 4 (6.2%), meanwhile mild PHT (25 - 40 mm Hg) in 3 (4.6%) of cases.

As for complications detected among our patients; most of the cases had recurrent chest infections in 24 (36.9%), followed by failure to thrive in 17 (26.2%), then heart failure in 17 (26.2%), pulmonary hypertension in 14 (21.5%) of them 3 patients (4.6%) developed reversal of the shunt and permanent cyanosis (Eisenmenger syndrome), and the less frequent were
Arrhythmia and pericardial effusion 1 (1.5%) per each, however no cases were detected with infective endocarditis or paradoxical embolization.

Concerning Relation between the type of ASD and occurrence of severe PHT, $P$-value = 0.02 (significant) revealing high association of severe PHT with primum ASD, severe PHT was detected among 4 patients; 3 of them had primum ASD those who developed reversal of the shunt and Eisenmenger Syndrome (cyanosis), while 1 patient had a large ASD II.

Concerning therapeutic modalities at time of the study; we found that, 38 (58.5%) of our patients needed specific cardiac medications, in the form of (anti-failure measures, inotropes, SBE prophylaxis, magnesium sulfate infusion and or oral sildenafil for treatment of PHT ......... etc). Therapeutic cardiac catheterization (percutaneous transcatheter device closure), however, was proceeded in 15 (23.1%) of cases predominantly ASDII type, while surgical closure was proceeded in 7 (10.8%) predominantly ASDI & sinus venosus types.

**Table (1): Age categories of studied cases at the time of presentation.**

<table>
<thead>
<tr>
<th>Age category</th>
<th>no</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neonates</td>
<td>13</td>
<td>20</td>
</tr>
<tr>
<td>Infants</td>
<td>32</td>
<td>49.2</td>
</tr>
<tr>
<td>Pre-school age</td>
<td>9</td>
<td>13.8</td>
</tr>
<tr>
<td>School-age</td>
<td>9</td>
<td>13.8</td>
</tr>
<tr>
<td>Adolescents</td>
<td>2</td>
<td>3.1</td>
</tr>
<tr>
<td>Total</td>
<td>65</td>
<td>100</td>
</tr>
</tbody>
</table>

ASD assessment was performed using transthoracic echocardiography (TTE) and or transoesophageal echocardiography (TEE).
Clinical Profile Of Atrial Septal Defects In children

Doaa Maghraby Bahig

Table (2): Echocardiographic findings among studied cases of ASD.

<table>
<thead>
<tr>
<th>Echo findings</th>
<th>no</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Type of ASD</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Primum type (ASDI)</td>
<td>7</td>
<td>10.8</td>
</tr>
<tr>
<td>Secundum type (ASDII)</td>
<td>53</td>
<td>81.5</td>
</tr>
<tr>
<td>Sinus venosus type</td>
<td>2</td>
<td>3.1</td>
</tr>
<tr>
<td>Combined</td>
<td>3</td>
<td>4.6</td>
</tr>
<tr>
<td>Size of ASD</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Small-sized (2-5 mm)</td>
<td>26</td>
<td>40</td>
</tr>
<tr>
<td>Medium-sized (5-8 mm)</td>
<td>18</td>
<td>27.7</td>
</tr>
<tr>
<td>Large sized (&gt;8 mm)</td>
<td>21</td>
<td>32.3</td>
</tr>
<tr>
<td>The direction of the shunt</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Left - to - right shunt</td>
<td>62</td>
<td>95.4</td>
</tr>
<tr>
<td>Right - to - left</td>
<td>3</td>
<td>4.6</td>
</tr>
<tr>
<td>Bidirectional</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Pulmonary blood pressure</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Normal (0-20 mmHg)</td>
<td>51</td>
<td>78.5</td>
</tr>
<tr>
<td>Increased (&gt;25 mmHg)</td>
<td>14</td>
<td>21.5</td>
</tr>
</tbody>
</table>

Discussion:

Defects in the atrial septum (ASDs) are one of the most common types of congenital heart defects (CHDs) in children and such a defect is the most common CHD in adults, they cause left to right shunt because the left atrial pressure is higher than that in the right atrium. This causes volume overloading of the right ventricle. The major types of ASD are Secundum, primum and sinus venosus types. (P.Syamasundar Rao, 2012)

Atrial Septal Defects of secundum type are generally well tolerated and usually do not cause symptoms during infancy and early childhood Bloch M, 2012, while those of primum or sinus venosus types usually cause early symptoms as poor physical development (failure to thrive), recurrent chest infections, poor feeding .......... etc. Abraham M. Rudolph, 2009. Development of exercise intolerance and arrhythmias in later childhood, adolescence, and adulthood, and the risk for development of pulmonary vascular obstructive disease in adulthood make these defects important. (P.Syamasundar Rao, 2012)

In this study, there was female predominance among total cases of ASD with male to female ratio (1:1.6), this was in agreement with recent studies as Osman Başpınar et al, 2006 in Turkey, Viralam S. et al, 2011 in India and Sabry A et al, 2010 at Benha University. In this work, there were gender differences among different morphologic types of ASD. For instance, ASDI was slightly more common in males, ASDII was more common in females with male to female ratio [1:2], whereas gender distribution was equal among cases with sinus venosus. Similar results had been conducted by Reller et al, 2008 in Metropolitan Atlanta and Andreas Hanslik et al, 2006 in Vienna.

In current study, we found that most of our studied cases (49.2%) presented during infancy, followed by (20%) during neonatal period, then (13.8%) during school age, (13.8%) also during pre-school age, and the less common was (3.1%) of patients presented during adolescence. These results correlate with Osman Başpınar et al, 2006 in Turkey who stated that (22.1%) of patients were newborns, (50%) were infants and
toddlers, (10.8%) were preschool children, (14.4%) were school children, while (2.7%) of patients were adolescents, whereas Sabry A et al, 2010 in Benha University stated age at presentation as follow, (19.3%) were neonates, (65.4%) were infants, whereas (15.4%) presented during preschool age. The declining age at diagnosis predates the introduction of echocardiography and may be attributed to improvements in social and medical circumstances over the period under study.

In our study, the commonest detected risk factor among studied cases was positive consanguinity, it was positive among (76.9%) of our cases, 1st degree consanguinity was detected among (64.6%) of patients, whereas (23.1%) had no consanguinity with a statistically significant difference between consanguineous and non-consanguineous patients (P-value = 0.01). When comparing these findings with other results, our results are highly in concordance with Al-Ani ZR, 2010 in Al-Ramadi, Iraq, who reported that Consanguinity was found in (78%) of cases with ASD; first cousin consanguinity comprised (66.2%) of cases, likewise, a study performed by Ramegowda et al, 2006 in South India, where he concluded that ASD is strongly influenced by consanguinity. The increased rate of consanguinity among parents of our patients is due to social and traditional habits concerned with marriage in Upper Egypt.

In the present work, we found that (47.6%) of patients were asymptomatic and discovered accidentally during routine medical examination, while (52.3%) were symptomatic. Symptoms included, recurrent chest infections in (36.9%) of patients the commonest, followed by respiratory distress in (32.3%), feeding difficulties in (30.8%), exertional dyspnea in (27.7%), failure to thrive in (26.2%), then manifestations of heart failure in (24.6%), whereas the less common was permanent cyanosis in 3 cases (4.6%), those who developed severe PHT with subsequent reversal of the shunt direction i.e. Eisenmenger syndrome. This was relatively in concordance with a study performed by Cardenas et al, 2007, who stated that the symptoms of ASD in studied children were recurrent respiratory infections in (36.5%) of the patients, failure to thrive or feeding problems in (5.7%), and cyanosis in (3.8%). Likewise, Edwin Francis et al, 2012 in records of 56 patients found that the commonest symptoms were failure to thrive in (75.1%), recurrent respiratory infections in (39.4%) and or heart failure in (7.2%) of studied patients. It has also been observed that symptoms (such as a failure to thrive) in the presence of an ASD in pediatric population, were more likely to be caused by extracardiac pathology, as reported by Andrews R et al, 2002, so, on finding an ASD in infants with failure to thrive, the clinician should continue the search for a cause of poor weight gain.

The high incidence of recurrent chest infections and failure to thrive among our patients in the present study could be explained by many reasons as poor ventilation, poor nutrition and low socioeconomic status especially in overcrowded and rural areas, also parasitic infestations, bad hygiene and improper dietary habits may contribute.

Echocardiography is the primary imaging modality used in the evaluation of ASDs because of its high sensitivity and specificity, low cost and easy accessibility. (Wang ZJ et al, 2003)

Concerning the frequency of different ASD morphologic types in our study, echocardiographic findings
demonstrated that the majority of cases were ASDII (81.5%), followed by ASDI in (10.8%), then combined ASD in (4.6%), and sinus venosus type the less common in (3.1%). These findings correlate with a study done by Jawad Khadim et al, 2009 in Basra, that included 68 patients with ASD, their ages ranged from 3 days to 19 years, most of them (80.9%) were of second type, (10.3%) were primum type, while rare types as sinus venosus represented (7.4%) of patients. In another study in the same regard, Sora Goetschmann et al, 2008 in Switzerland studied 355 pediatric patients, of them (83.6%) were ASDII, whereas (16.3%) were ASDI, however, John J et al, 2011 reported that Mixed defects accounted for only (7%) of all cases with atrial septal defects.

In our study, the size of the defect among total cases of different anatomical types of ASD as measured by ECHO, ranged from (1.5 - 23 mm) with mean ± SD (7.9 ± 4.7), small defects (2 - 5 mm) were detected in (40%) of cases, medium (5 - 8 mm) in (27.7%), while large-sized defects (>8 mm) were detected in (32.3%) of cases. As for the size of the defect among cases in relationship to anatomical types of ASD, patients with secundum ASD for example, had small-sized defect in (47.2%) of cases, the commonest, followed by medium-sized in (28.3%), and the less common was a large-sized defect in (24.5%). These findings were in concordance with Andreas Hanslik et al, 2006 in Vienna, where he studied Predictors of Spontaneous Closure of Isolated Secundum Atrial Septal Defect in Children. The median age at diagnosis of ASDII was (0 - 13.9 years), the atrial septal defect diameter at diagnosis was 4 to 5 mm in (40%) of cases, 6 to 7 mm in (28%), 8 to 10 mm in (21%), and > 10 mm in (11%).

As for direction of the shunt; the vast majority of cases (95.4%) had left to right shunt, while 3 cases (4.6%) had right to left shunt [3 cases with primum ASD of them 1 case had down syndrome] those who developed severe PHT with the subsequent reversal of the shunt i.e. Eisenmenger syndrome. Similar results had been conducted by Awasthy N. et al, 2013 in Delhi, India, where he stated that Left to right shunt, in associated ASD without pulmonary artery hypertension (PAH) this is the commonest pattern of shunting. While right to left shunting will occur in severe PHT.

In our work, Pulmonary hypertension (PHT) was confirmed and measured by ECHO; severe PHT was detected in 4 cases representing (6.2%) of patients (1 case had a large secundum ASD, while 3 cases had large primum ASDs of them 1 case had Down syndrome). Several other investigators, however, reported the following results, GVaksmann et al, 2009 in France for instance, reported 9 cases of Atrial Septal Defect with severe pulmonary hypertension (5 cases of them had primum ASDs, while 4 cases had secundum ASDs), these cases represent (3.5%) of the 255 cases of Atrial Septal Defect seen at the Hospital Cardiologique of Lille. The relatively high incidence of severe PHT among our studied children compared with other studies could be contributed to the small sample size of our studied patients.

As for complications recorded in this work, most of the cases had recurrent chest infections in (36.9%), followed by failure to thrive in (26.2%), then heart failure in (26.2%), pulmonary hypertension in (21.5%) of them (4.6%) developed permanent cyanosis (Eisenmenger syndrome), and the less frequent were arrhythmia and pericardial effusion (PE) each of them represented (1.5%), while, no cases
were recorded of infective endocarditis or paradoxical embolization. As for pericardial effusion (PE), it had been detected in one female patient who had a large secundum ASD, these results are largely consistent with the study conducted by Surendranath Reddy Veeram Reddy et al, 2009 in Michigan, where he demonstrated that the prevalence of global pericardial effusion in children with isolated secundum ASD was rare (1.1%), but they had significantly more pericardial effusions compared with children with normal hearts, Allen HD et al, 2008 as well, reported that Patients with septal defects leading to excessive pulmonary blood flow are at an increased risk of pulmonary infections and para-pneumonic pleural and pericardial effusions. Several other investigators, however, reported contradictory results, for instance C. Osborne Shuler et al, 2013 in Columbia, in a pediatric cohort study among children with ASD, recorded complications in the form of pulmonary hypertension (3.8%), and arrhythmia in the form of supraventricular tachycardia (2.4%), also Nawal Azhari et al, 2004 in Saudi Arabia, showed that (41.3%) of studied ASD patients had failed to thrive, and (11.6%) had congestive heart failure.

Concerning therapeutic modalities at time of the study; we found that, (58.5%) of our patients needed specific cardiac medications in the form of (anti-failure measures, inotropes, SBE prophylaxis, magnesium sulfate infusion and or oral sildenafil for treatment of PHT......etc), however interventional cardiac catheterization (device closure) was proceeded in (23.1%) of cases predominantly of ASD secundum type, while (10.8%) of patients underwent surgical closure those predominantly with ASD primum and sinus venosus types. Similar results had been conducted by C.Osborne Shuler et al, 2013 who stated that Pharmacologic treatments, predominantly diuretics, were prescribed for 21% of the cohort. Surgical closures (6.3%) and transcatheter closures (1.4%) were used for ASD secundum cases, whereas surgical closures predominated for ASD primum (25.6%) and sinus venosus (13.5%) lesions.

In our study, a significant correlation was found between the type of ASD and frequency of severe PHT, (p-value = 0.02), revealing high association of severe PHT with primum ASD, severe PHT was detected among 4 patients (as mentioned before); 3 of them had primum ASD those who developed reversal of the direction of the shunt and Eisenmenger Syndrome (cyanosis), while 1 patient had a large ASD II. These findings come in agreement with a study done by G Vaksmann et al, 2009 in France, where he reported 9 cases of Atrial Septal Defect with severe pulmonary hypertension (5 cases of them had primum ASDs, while 4 cases had secundum ASDs), these cases represent (3.5%) of the 255 cases of Atrial Septal Defect seen at the Hospital Cardiologique of Lille.

Conclusions:
The great majority of infants with ASDII are most often asymptomatic. Frequently, the condition is detected by a murmur on routine physical examination when they are school-aged children. Even a large ASD rarely produces clinically evident heart failure in childhood. In younger children, subtle failure to thrive may be present, in older children, varying degrees of exercise intolerance may be noted. The predominance of small-sized defects in our study is explained...
by the frequent use of echocardiography for infants for various indications, for example, in NICUs and for the evaluation of heart murmurs. Echocardiography may result in early discovery of small defects in infants that otherwise might not have been diagnosed at all. While in older children, small defects have already closed spontaneously, leaving mainly large and medium-sized defects to be detected.

References


